FOXP2 gene

forkhead box P2

Normal Function

The *FOXP2* gene provides instructions for making a protein called forkhead box P2. This protein is a transcription factor, which means that it controls the activity of other genes. It attaches (binds) to the DNA of these genes through a region known as a forkhead domain. Researchers suspect that the forkhead box P2 protein may regulate hundreds of genes, although only some of its targets have been identified.

The forkhead box P2 protein is active in several tissues, including the brain, both before and after birth. Studies suggest that it plays important roles in brain development, including the growth of nerve cells (neurons) and the transmission of signals between them. It is also involved in synaptic plasticity, which is the ability of connections between neurons (synapses) to change and adapt to experience over time. Synaptic plasticity is necessary for learning and memory.

The forkhead box P2 protein appears to be essential for the normal development of speech and language. Researchers are working to identify the genes regulated by forkhead box P2 that are critical for learning these skills.

Health Conditions Related to Genetic Changes

FOXP2-related speech and language disorder

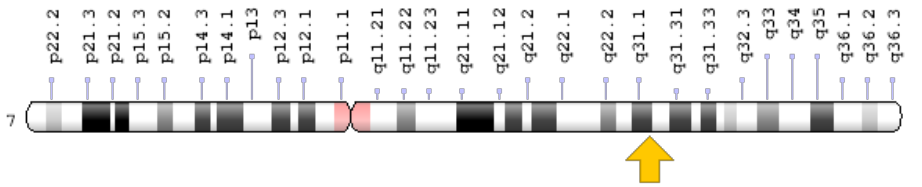
Several changes involving the *FOXP2* gene can result in *FOXP2*-related speech and language disorder, a condition that affects the development of speech and language starting in early childhood. Some affected individuals have a deletion that removes a small segment of chromosome 7, including the *FOXP2* gene and several neighboring genes. Other people with this condition have a mutation within the *FOXP2* gene itself. Less commonly, *FOXP2*-related speech and language disorder results from a rearrangement of the structure of chromosome 7 (such as a translocation) or from inheriting two copies of chromosome 7 from the mother instead of one from each parent (a phenomenon called maternal uniparental disomy or maternal UPD). It remains unclear how having two maternal copies of chromosome 7 affects the activity of the *FOXP2* gene.

The genetic changes that underlie *FOXP2*-related speech and language disorder disrupt the activity of the *FOXP2* gene. Because forkhead box P2 is a transcription factor, these changes affect the activity of other genes in the developing brain. Researchers are working to determine which of these genes are involved and how changes in their activity lead to abnormal speech and language development.

Chromosomal Location

Cytogenetic Location: 7q31.1, which is the long (q) arm of chromosome 7 at position 31.1

Molecular Location: base pairs 114,086,310 to 114,693,772 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CAG repeat protein 44
- CAGH44
- forkhead/winged-helix transcription factor
- SPCH1
- TNRC10
- trinucleotide repeat containing 10

Additional Information & Resources

Educational Resources

- Coffee Break: Tutorials for NCBI Tools (2003): Talking About the Genetics of Talking
  https://www.ncbi.nlm.nih.gov/books/NBK2338/
- Madame Curie Bioscience Database (2009): FOXP Genes, Neural Development, Speech and Language Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK7023/

GeneReviews

- FOXP2-Related Speech and Language Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK368474

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FOXP2%5BTIAB%5D%29+OR+%28forkhead+box+P2%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- FORKHEAD BOX P2
  http://omim.org/entry/605317

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_FOXP2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FOXP2%5Bgene%5D
- HGNC Gene Family: Forkhead boxes
  http://www.genenames.org/cgi-bin/genefamilies/set/508
- HGNC Gene Family: Trinucleotide repeat containing
  http://www.genenames.org/cgi-bin/genefamilies/set/775
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=13875
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/93986
- UniProt
  http://www.uniprot.org/uniprot/O15409

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